Exhibit A

Precoordination of diagnosis concepts for increased granularity in a clinical genomic ontology

Abstract

Clinical Genomics is an emerging discipline that interconnects phenotypic and genomic information to accelerate research and development of new medical therapies. Critical informatics needs of clinical genomic researchers are the ability to identify clinical concepts at an appropriate level of granularity and the ability to query with specific values of these concepts. The clinical diagnosis is the data element of greatest importance to these researchers. To represent and communicate diagnosis concepts, their definitions are precoordinated at the desired granularity. Names and codes for these complex concepts are constructed from simpler, 'building block' clinical concepts. Processes are defined to review and remedy user-detected gaps in the ontology. The precoordinated diagnoses provide genomic researchers the required granularity and specificity without the burden of inappropriate diagnosis constructs being introduced into the database.